



NEWBORN SCREENING *News*

SUMMER 2005

THE CALIFORNIA NEWBORN SCREENING PROGRAM

NBS Program to Expand Summer 2005

As a result of Senate Bill 142 legislation, screening for congenital adrenal hyperplasia (CAH) and disorders detectable by tandem mass spectrometry (MS/MS) will begin by August 1, 2005. In addition to adding CAH and multiple metabolic disorders to the panel of detectable conditions, there will be a number of substantial changes made to the California Newborn Screening Program starting this summer. Some of these include:

- A new web-based computer system,
- New specimen collection and other NBS forms,
- New and revised educational materials for health care providers and parents, and
- A change in the retesting procedure for newborns that do not have an adequate pre-transfusion specimen.

Disorders to be Added by August 2005

Congenital Adrenal Hyperplasia

The Program will screen for two forms of classical congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency:

- Salt-Wasting CAH
- Simple Virilizing CAH

In addition, some newborns with non-classical congenital adrenal hyperplasia (NCAH), a less severe form of the disorder, will be detected.

Multiple Metabolic Conditions

The NBS Program will be able to detect via tandem mass spectrometry (MS/MS) over 40 additional conditions including:

- Amino Acid Disorders
- Organic Acid Disorders
- Fatty Acid Oxidation Disorders

Expected Number of Newborns to be Detected with these Disorders

When the Program's new expansion is implemented, we anticipate detecting 115-150 newborns every year with one of these added disorders. Specifically, we expect to detect:

- 35-50 newborns with classical congenital adrenal hyperplasia
- 80-100 newborns with a metabolic disorder other than PKU

These estimates are based upon the findings of other states that have been testing for these conditions and the results from our 18 month MS/MS pilot project. With these additions, we anticipate identifying over 625 newborns each year with a clinically significant condition, most of who will benefit from early diagnosis and treatment. Benefits include prevention of: death, mental retardation, or severe health problems associated with absence or delay of treatment.

Why Are these Disorders being Added?

Legislation authored by Senator Alpert and signed by the Governor has authorized the expansion of the Newborn Screening Program to include these disorders by August 1, 2005. These changes are consistent with national recommendations being developed by the American College of Medical Genetics (ACMG), with funding from the Health Resources and Services Administration (HRSA), which strive for a uniform newborn screening panel and system throughout the country. ACMG's recommendations and the expansion in California are in response to the availability of new screening technology and advances in the diagnosis and treatment of clinically significant disorders identifiable at birth. Other professional and parent groups have supported this effort.

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Description of Disorders

Congenital Adrenal Hyperplasia

In both types of classical CAH the adrenal glands do not make enough of the stress-fighting hormone cortisol. In about two-thirds of the cases, newborns also do not produce enough of the salt-retaining hormone aldosterone which can lead to dehydration, shock, and even death. Treatment with one or more oral medications can prevent these problems. Girls with CAH may have the additional problem of masculine-looking external genitals, which may later need to be corrected with surgery.

Metabolic Disorders

Metabolic disorders affect the body's ability to use amino acids (from proteins), organic acids (from proteins, fats, and sugars) or fatty acids (from fats) all of which are necessary for energy, growth and repair.

To breakdown or convert these substances, certain enzymes must be present. When there is a deficiency or absence of the needed enzyme (i.e., a block in the metabolic pathway), metabolites build up in large amounts and may be harmful to the body. Metabolic conditions have varying degrees of severity. If identified early, many of these disorders can be treated before they cause serious health problems. Without treatment, newborns can become ill very quickly and die. Treatment may include close monitoring of the child's health, medication, dietary supplements, and/or special diets including medical formulas and foods.

Screening Methodology

Amino Acid, Organic Acid and Fatty Acid Disorders:

Tandem Mass Spectrometry allows for the detection of over 40 disorders using a single analytical run. Forty-

two analytes are measured for each newborn. A positive screening report for one or more conditions will result from: 1) Elevations in analytes and/or ratios of certain analytes, or 2) specific patterns of analytes consistent with a known disorder. For a newborn with a positive screening result, the regional NBS Area Service Center (ASC) coordinator will telephone the newborn's primary care provider, provide information about the possible disorder(s) and assist with an immediate referral to a California Children's Services (CCS)-approved Metabolic Center for a diagnostic evaluation.

Congenital Adrenal Hyperplasia:

Screening for CAH will consist of two-tier testing. The first tier measures 17-hydroxyprogesterone (17-OHP) using the AutoDelphia instrument (the same machine that measures TSH for primary congenital hypothyroidism screening). This is an immunofluorescence-assay (FIA). 17-OHP is elevated in newborns with classical CAH due to the pathway to cortisol being blocked by the absence of the enzyme, 21-hydroxylase. Newborns with very high first tier 17-OHP values, based upon four birth weight categories, will be referred immediately to a CCS-approved Endocrine Center for a diagnostic evaluation.

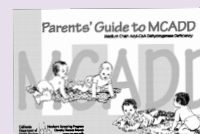
The second tier test will be performed if a newborn has values that are elevated but below the initial cut-offs for immediate reporting. The second test will be performed on the initial blood specimen via Tandem Mass Spectrometry. This test will measure 17-OHP, androstenedione and cortisol. Newborns with an elevated 17-OHP and a high ratio of 17-OHP and androstenedione to cortisol will be referred to a CCS-approved Endocrine Center or CCS-paneled pediatric

endocrinologist for a diagnostic evaluation.

Newborns with non-classical congenital adrenal hyperplasia (NCAH) have only slightly elevated 17-OHP values and, therefore, the test will only identify some of the newborns with NCAH.

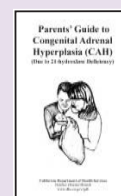
Additional Information for Health Care Providers and Families

The NBS program website is being updated to include more detailed information for both providers and parents on the disorders being added and other changes that will occur this summer. Fact sheets and educational materials for most of the conditions detectable via MS/MS screening will be available for primary care providers and parents from the NBS Area Service Centers, and State NBS office at (510) 412-1542.



These materials can also be downloaded from the NBS website. Recent

Program newsletters and letters to hospitals and providers are also posted on our website.



A Parent's Guide to Congenital Adrenal Hyperplasia will also be available for distribution to families.

New Specimen Collection Form

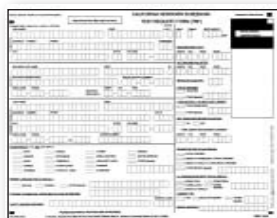
As part of the NBS expansion, the State will implement a new web-based computer system, referred to as the Screening Information System (SIS), that utilizes optical character recognition (i.e., scanning) and an automated filter paper card punching system. To accommodate these major program changes we have made substantial changes to the newborn screening specimen collection form. The current form, which consists of the demographic form and the filter paper on which the blood is collected, will be replaced by two separate forms - the test request form (TRF) on which demographic data is captured and the specimen collection card (filter paper). **To guard against errors and improve the efficiency of newborn screening, the person completing the form will need to print legibly in capital letters, using only black or dark blue ink.**

The two forms will be linked using barcodes. Once a specimen is collected, the two barcode stickers on the specimen collection card will be peeled off. One will be affixed to the original white TRF and the other to the goldenrod copy of the TRF, which is filed in the newborn's medical record. Do not fold the original copy of the TRF. (This interferes with the scanning process.) The forms should then be sent together to the assigned Newborn and Prenatal Screening (NAPS) laboratory.

Hospitals and other newborn screening providers should have received sample copies of the new NBS TRFs and specimen collection cards, along with a poster and handouts on how to collect NBS specimens using the new TRF and card. Please follow the procedures on the poster exactly in order to avoid

misidentification of specimens.

If you have not received these, want additional materials or would like assistance with in-service training of your staff, call your assigned Area Service Center.



Test request form



Specimen collection card

Advantages of the new TRF/ Specimen Collection Card

- It is easier to hold the new filter paper card for collection.
- Less room is needed for drying the specimen.
- Separate specimen collection cards allow for use of an automated puncher at the screening laboratories.
- If an error is made in completing the TRF prior to applying the bar code strips, a new one can be used without having to collect another specimen from the baby.
- The new TRF will be scanned, which should provide for increased accuracy of information entered into the State computer system.
- Hospitals have the option of using their own computer systems to print the demographic information from the newborn's computer medical record directly onto the TRF, provided that the information is printed inside the red boxes. Hospitals will still be required to use the State TRF, so the TRF will need to be fed into the printer. Regular paper is not acceptable and the

information will need to be lined up correctly with the boxes on the form. The specimen collection information (e.g., collection date, time, and collector's initials) will still need to be done by hand at the time of collection.

- Separating the filter paper from the TRF allows for use of less filter paper, which is the most expensive part of the form. This enables the Program to keep the cost to hospitals at \$1.00 per set (TRF and specimen collection card).
- Separating the TRF from the blood collection card paves the way for future possibilities, such as collaborative data collection and electronic data transfer.

Credit for Discontinued Specimen Collection Forms

Supplies of new TRFs and specimen collection cards will be sent to all hospitals in the near future. After receipt of the new forms, full (but not opened) packs of the old forms can be returned to the State for a credit of \$1.00 per form. Partial packs of the old form should be used up as soon as possible. Hospitals will receive specific instructions on returning the old forms.

Other New NBS Forms and Reports

The NBS-OH, NBS-MR, and NBS-NO forms have also been revised to allow for optical character recognition (OCR) scanning of information into the State computer system. Supplies were sent out beginning in March 2005. For additional copies of the forms call: (510) 412-3950. The Newborn Screening Result Mailers and the Hospital Evaluation Performance Profile (HEPP) report will be modified to accommodate the changes to the Program.

Change in Follow-up for Transfused Babies

Transfused infants who do not have an adequate pre-transfusion specimen will be required to have another filter paper specimen drawn at least 24 hours post-transfusion to test for metabolic (except galactosemia) and endocrine disorders, and a whole blood specimen collected to screen for hemoglobinopathies.

While metabolic and endocrine screening results are considered valid 24 hours after a red blood cell transfusion, hemoglobinopathy and galactosemia screening results are invalid for at least three months. Although physicians have had the option of having babies who did not have an adequate pre-transfusion test result retested for hemoglobinopathies in three months, few have done so through the Program. In view of the significant prevalence of hemoglobinopathies in California's general population (1:3,000), the Program now requires that babies who do not have an adequate pre-transfusion screening specimen be promptly tested for hemoglobinopathies utilizing DNA analysis of white blood cells.

This testing, offered through the NBS Program, can be performed right away, permitting early detection and treatment of babies with hemoglobinopathies who were transfused prior to screening. A whole blood specimen will need to be collected and sent to the NBS Hemoglobin Reference Laboratory at Children's Hospital and Research Center, Oakland. There is no charge for this service. ASC staff will contact primary care providers and provide the specific instructions to assure this additional testing is done when needed.

In these cases, DNA testing will detect the following: S, C, D, E, Knossos, and O Arab hemoglobin patterns, and the most common beta globin mutations that cause beta thalassemia. Additionally, those transfused babies with at least one Asian parent will have DNA testing for the seven most common alpha thalassemia deletions, which are found almost exclusively in individuals of Asian descent. For purposes of the follow-up, Asian is considered to be any of the ethnicities listed on the specimen collection form except Black, Middle Eastern, Native American, Hispanic or White.

The galactosemia results on post-transfusion specimens will also be invalid. Follow-up testing for galactosemia will not be routinely required. However, if a baby has symptoms of galactosemia (see table to right) or if there is a family history of the disorder, immediate parent testing for carrier status is strongly recommended. Immediately contact the ASC for assistance. Parent testing is done, at no charge, through the State Galactosemia Confirmatory Laboratory at Children's Hospital, Los Angeles (CHLA).

Timing of Specimen Collection

The timing of specimen collection is one of the key factors in the assurance of test validity and reliability. Because of the initial instability of a neonate's metabolic system, screening results of a specimen collected in the first hours of life may not be reflective of the newborn's true status.

The addition of the multiple metabolic conditions and CAH does not change the NBS Program's requirement for the timing of specimen collection. Specimens should be collected on newborns after 12 hours of age, **as close to discharge as possible**, but no later than the sixth day of age. The specimen should always be collected prior to the first red cell transfusion, regardless of the age of the newborn. If the newborn is less than 12 hours of age at collection, a second specimen will be required after 12 hours of age but no later than 6 days of age.

GALACTOSEMIA

Symptoms May Include:

- Prolonged jaundice
- Cataracts
- Lethargy and poor feeding
- Susceptibility to infection

Urine Dipstick Results:

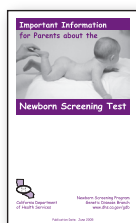
1. A positive Clinitest for reducing substances indicates either glucose or galactose elevation, followed by
2. A negative test using a glucose oxidase strip like Clinistix rules out glucose and indicates galactose elevation

Review of the Roles of the Key Providers in Newborn Screening

Prenatal care providers, hospital staff and pediatric care providers all have screening responsibilities mandated by California law. The following is a summary of the key responsibilities as outlined in Program regulations and guidelines.

Prenatal Care Provider

- ☑ **Provide parents with a copy of Important Information for Parents about the Newborn Screening Test (IIP).** State law



requires that prenatal care providers distribute this booklet to all pregnant women under their care prior to the woman's estimated date of delivery. A new version will be distributed for use when the Program expands.

Copies of these booklets will be sent out with the Expanded AFP (X-AFP) Screening Program supply orders; however, they will no longer be put in an envelope with the X-AFP Program booklet. Copies of IIP can also be obtained free of charge from the State by calling (510) 412-1542. Studies have shown that the ideal time to give this information to expecting couples is during the third trimester either as part of a birthing class or at a routine office visit. They are least effective when given as part of a large packet of materials.

- ☑ **Answer any questions of expectant parents about newborn screening.**

- ☑ If there is a family history of a disorder not included in the NBS Program, **discuss diagnostic testing at a private laboratory, when available, with parents.**

Hospital Staff (during hospital stay for delivery)

- ☑ **Provide parents with a copy of Important Information for Parents about the Newborn Screening Test (IIP).**

- ☑ **Complete NBS Test Request Form.** All information must be legible and accurate including contact information for the parent and the pediatric care provider who will be providing well baby care of the infant (medical home).

- ☑ **Follow State guidelines on specimen collection and handling.** (Written instructions are attached to the filter paper card, or available on our website.)

For information or a copy of the National Committee for Clinical Laboratory Standards' (NCCLS) video Making a Difference Through Newborn Screening: Blood Collection on Filter Paper-California version, and/or the NBS Program poster and handout on specimen collection using the new test request form and filter paper cards, call your assigned regional NBS Area Service Center.

- ☑ **Match birth logs/records with newborn screening forms** to ensure that a specimen is collected on each newborn prior to discharge.

- ☑ **Notify the State of any newborn that was discharged without having a specimen collected** using the Hospital Report of Newborn Specimen Not Obtained (NBS-NO) form provided by the State.

- ☑ **Check the medical record of all newborns** at 14 days after discharge for a copy of the newborn screening test results to ensure the specimen was received by and tested at the laboratory. Notify the State using the *Provider Request for Missing Newborn Screening Test Results* (NBS-MR) form if it is missing.

Pediatric Care Provider

- ☑ **Review the newborn screening results of all new patients under one year of age to ensure testing and follow-up has been provided.¹**

Results will be automatically sent to the hospital of birth and pediatric care provider as listed by the hospital on the NBS test request form. Copies of the results can be obtained by contacting the State office or local NBS Area Service Center. HIPAA requires providers who are not listed on the NBS Test Request Form to fax a parent release or parent consent to treat form to GDB at (510) 412-1559 to receive the results.

- ☑ **Inform parents of the results of the NBS test** and ensure appropriate follow-up is provided for any newborn with a non-negative test result. ASC staff will inform pediatric care providers by phone of the NBS results for newborns with initial inadequate or positive test results and assist them in the notification of parents, arranging for retesting, and/or confirmatory testing. ASC staff can also provide information on the disorders or arrange for a medical sub-specialist (metabolic, endocrine or hematology) to contact the primary care providers to discuss recommended diagnostic and treatment services.

- ☑ **Refer newborns with positive results to CCS-approved Special Care Centers** or CCS-paneled sub-specialists for the initial diagnostic

¹ Pediatrics 89(2), February 1992

evaluation and treatment when appropriate per NBS Program recommendations. ASC staff will assist with the referral process, including notifying the local CCS county office to obtain CCS authorization.

☑ **Remain watchful** of any signs or symptoms of these diseases in your patients. The possibility of a disorder should not be ruled out solely on the basis of the newborn screening results.

Referral for Diagnostic Evaluation and Treatment

All newborns identified with a disorder through the NBS Program should have access to a diagnostic evaluation through a CCS-approved Special Care Center (SCC). Specialists at the SCC will work closely with the primary care provider in determining what testing is needed and in the development of a treatment plan when necessary. When a disorder is confirmed, the NBS Program strongly recommends that newborns receive ongoing specialty care at a SCC where a multi-disciplinary team (physicians, dietician, nurse, social worker, genetic counselor) can provide a comprehensive approach to assisting the family.

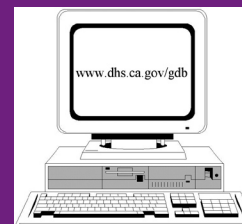
Who Will Pay for the Diagnostic Evaluation and Treatment if Needed?

All newborns referred to a CCS-approved Special Care Center (SCC) by the California Newborn Screening Program are eligible for a diagnostic evaluation through the SCC regardless of income. The ASC will work with the primary care provider in determining which SCC to refer the baby to based upon location and insurance plan coverage. Because the disorders screened for by the NBS Program require immediate follow-up, the CCS program has developed an expedited authorization process for the initial diagnostic evaluation at a SCC.

Parents will be asked to complete an application form to determine eligibility for CCS payment for the diagnostic evaluation. Most health insurance and health maintenance organizations (HMOs) provide at least some coverage for the diagnostic evaluation and any necessary treatment. If a baby has health insurance the SCC will bill the health insurance company or HMO for the services. Infants who have full scope, no share of cost Medi-Cal coverage, or are Healthy Families subscribers will be authorized by CCS for diagnostic and treatment services and parents will not need to pay anything for services. If parents do not have health insurance, or their insurance only covers partial payment, the infant may be eligible for the CCS program. Eligibility for coverage of treatment costs through the CCS program is based on having a CCS-eligible medical condition, and meeting program financial and residential eligibility. For more information on the CCS program visit their website at: <http://www.dhs.ca.gov/pcfh/cms/ccs/>. For a list of SCCs see page 7 of this newsletter.

Limitations of the Newborn Screening Program

Due to biological variability of newborns and differences in detection rates for the various disorders in the newborn period, the Newborn Screening Program will not identify all newborns with these conditions. While a positive screening result identifies newborns at an increased risk to justify a diagnostic work-up, a negative screening result does not rule out the possibility of a disorder. Health care providers should remain watchful for any signs or symptoms of these disorders in their patients. **A newborn screening result should not be considered diagnostic, and cannot replace the individualized evaluation and diagnosis of an infant by a well-trained, knowledgeable health care provider.**



For more information about the Newborn Screening Program please visit our website at: www.dhs.ca.gov/gdb then click on Newborn Screening.

CCS – Approved Metabolic Centers

Cedars-Sinai Medical Center

Los Angeles, CA 90048
(310) 423-9914

Children's Hospital & Research Center at Oakland

Oakland, CA 94609
(510) 428-3550

Children's Hospital Central California

Madera, CA 93638
(559) 353-6400

Children's Hospital of Los Angeles

Los Angeles, CA 90027
(323) 660-2450

Children's Hospital Orange County

Orange, CA 92668
(714) 532-8852

Children's Hospital & Health Center

San Diego, CA 92123
(619) 543-7800

Harbor/UCLA Medical Center

Torrance, CA 90509
(310) 222-3756

Kaiser Permanente Medical Center

Oakland, CA 94611
(510) 752-7703

Kaiser Permanente Southern California

Los Angeles, CA 90027
(323) 783-6970

Los Angeles County/USC Medical Center

Los Angeles, CA 90033
(323) 226-3816

Lucile Salter Packard Children's Hospital at Stanford

Palo Alto, CA 94301
(650) 723-6858

Sutter Medical Center

Sacramento, CA 95819
(916) 733-6023

UC Davis Medical Center

Sacramento, CA 95817
(916) 734-3112

UC San Francisco Medical Center

San Francisco, CA 94143
(415) 476-2757

UC Los Angeles Medical Center

Los Angeles, CA
(310) 206-6581

UC Irvine Medical Center

Orange, CA 92868
(714) 456-8513

CCS – Approved Endocrine Centers

Cedars-Sinai Medical Center

Los Angeles, CA 90048
(310) 423-7940

Children's Hospital – Oakland

Oakland, CA 94609
(510) 428-3654

Children's Hospital Central California

Madera, CA 93638
(559) 353-8700

Children's Hospital of Los Angeles

Los Angeles, CA 90027
(323) 660-2450

Children's Hospital of Orange County

Orange, CA 92668
(714) 532-8634

Children's Hospital & Health Center

San Diego, CA 92123
(858) 966-4032

Harbor/UCLA Medical Center

Torrance, CA 90509
(310) 222-2394

Loma Linda University Medical Center

Loma Linda, CA 92354
(909) 558-2827

Lucile Salter Packard Children's Hospital at Stanford

Palo Alto, CA 94304
(650) 723-5791

Miller Children's at Long Beach Memorial Medical Center

Long Beach, CA 90801
(562) 933-8562

Santa Clara Valley Medical Center

San Jose, CA 95128
(408) 885-5405

Sutter Memorial Hospital

Sacramento, CA 95819
(916) 733-6006

UC Davis Medical Center

Sacramento, CA 95817
(916) 734-3112

UC San Francisco Medical Center

San Francisco, CA 94143
(415) 476-1016

UC Los Angeles Medical Center

Los Angeles, CA 90095
(310) 825-6244

CCS – Approved Sickle Cell Disease/Hemoglobin Centers

Cedars-Sinai Medical Center

Los Angeles, CA 90048
(310) 423-4423

Children's Hospital & Research Center at Oakland

Oakland, CA 94609
(510) 428-3651

Children's Hospital Central California

Madera, CA 93638
(559) 353-5461

Children's Hospital of Los Angeles

Los Angeles, CA 90027
(323) 660-2450

Children's Hospital of Orange County

Orange, CA 92868
(714) 532-8459

Children's Hospital & Health Center

San Diego, CA 92123
(858) 966-5811

City of Hope Medical Center

Duarte, CA 91010
(626) 256-4673 ext. 62913

Harbor/UCLA Medical Center

Torrance, CA 90502
(310) 222-2394

Kaiser Permanente Medical Center

Oakland, CA 94611
(510) 752-6192

Kaiser Permanente Medical Center

West Los Angeles
Los Angeles, CA 90034
(323) 857-2000

Loma Linda University

Loma Linda, CA 92354
(909) 558-2617

Los Angeles County USC Medical Center

Los Angeles, CA 90033
(323) 226-3853

Lucile Salter Packard Children's Hospital at Stanford

Palo Alto, CA 94304
(650) 725-1072

Miller Children's at Long Beach Memorial Medical Center

Long Beach, CA 90801
(562) 492-1062

Saint Agnes Medical Center

Fresno, CA 93720
(209) 449-5121

Sutter CHS Central

Sacramento, CA 95816
(916) 733-1757

UC Davis Medical Center

Sacramento, CA 95817
(916) 734-2781

UC San Francisco

San Francisco, CA 94143
(415) 502-8034

UC Irvine

Orange, CA 92868
(714) 456-5680

UCLA Hospital & Clinics

Los Angeles, CA 90095
(310) 825-6708

Newborn Screening Area Service Centers (NBS-ASCs)

Stanford University
(650) 812-0353

Children's Hospital Central CA
(559) 353-6416

UCLA Medical Center
(310) 826-4458

Harbor/UCLA Medical Center
(310) 222-3751

University of California, San Diego
(858) 300-1081

Kaiser Permanente, Northern CA
(510) 752-6192

Kaiser Permanente, Southern CA
(626) 564-3322

Disorders Detectable by the California Newborn's Screening Program as of Summer 2005*

I. Metabolic Disorders

A. Carbohydrate Disorders

- classical galactosemia

B. Amino Acid Disorders

- classical phenylketonuria (PKU)
- variant PKU
- GTPCH deficiency (biopterin deficiency)
- PTPS deficiency (biopterin deficiency)
- DHPR deficiency (biopterin deficiency)
- PH deficiency (biopterin deficiency)
- argininemia/arginase deficiency
- argininosuccinic acid lyase deficiency (ASAL deficiency)
- citrullinemia, Type I/argininosuccinic acid synthetase deficiency (ASAS deficiency)
- citrullinemia, Type II (citrin deficiency)
- gyrate atrophy of the choroid and retina
- homocitrullinuria, hyperornithinemia, hyperammonemia – HHH
- homocystinuria/cystathionine beta-synthase deficiency (CBS deficiency)
- methionine adenosyltransferase deficiency (MAT deficiency)
- maple syrup urine disease (MSUD)
- non-ketotic hyperglycinemia
- prolinemia
- tyrosinemia

C. Organic Acid Disorders

- 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency
- 2-methylbutyryl-CoA dehydrogenase deficiency
- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMGCoA lyase deficiency)
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC deficiency)

- 3-methylglutaconic aciduria (MGA), Type I (3-methylglutaconyl-CoA hydratase deficiency)
- 5-oxoprolinuria
- beta-ketothiolase deficiency (BKT)
- ethylmalonic encephalopathy (EE)
- glutaric acidemia type-1 (GA-1)
- isobutyryl-CoA dehydrogenase deficiency
- isovaleric acidemia (IVA)
- malonic aciduria
- methylmalonic acidemia, mut –
- methylmalonic acidemia, mut O
- methylmalonic acidemia (Cbl A, B)
- methylmalonic acidemia (Cbl C, D)
- multiple carboxylase deficiency (MCD)
- propionic acidemia (PA)

D. Fatty Acid Oxidation Disorders

- carnitine transporter deficiency
- carnitine-acylcarnitine translocase deficiency (CAT deficiency)
- carnitine palmitoyl transferase deficiency-type 1 (CPT-1 deficiency)
- carnitine palmitoyl transferase deficiency-type 2 (CPT-2 deficiency)
- long chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD deficiency)
- medium chain acyl-CoA dehydrogenase deficiency (MCAD deficiency)
- multiple acyl-CoA dehydrogenase deficiency (MAD deficiency)/glutaric acidemia type-2 (GA-2)
- short chain acyl-CoA dehydrogenase deficiency (SCAD deficiency)
- trifunctional protein deficiency (TFP deficiency)

- very long chain acyl-CoA dehydrogenase deficiency (VLCAD deficiency)

II. Endocrine Disorders

- primary congenital hypothyroidism
- variant hypothyroidism
- congenital adrenal hyperplasia-salt wasting (21-hydroxylase deficiency)
- congenital adrenal hyperplasia-simple virilizing (21-hydroxylase deficiency)

III. Hemoglobin Disorders

- sickle cell anemia (Hb S/S disease)
- sickle C disease (Hb S/C disease)
- sickle D disease (Hb S/D disease)
- sickle E disease (Hb S/E disease)
- Hb S/hereditary persistence of fetal hemoglobin (Hb S/HPFH)
- sickle cell disease variant (other sickle cell disease, Hb S/V)
- Hb S/Beta⁰ thalassemia
- Hb S/Beta⁺ thalassemia
- Hb C disease (Hb CC)
- Hb D disease (Hb DD)
- alpha thalassemia major
- Hb H disease
- Hb H/Constant Spring disease
- beta thalassemia major
- Hb E/Beta⁰ thalassemia
- Hb E/Beta⁺ thalassemia
- Hb E/Delta Beta thalassemia
- Hb C/Beta⁰ thalassemia
- Hb C/Beta⁺ thalassemia
- Hb D/Beta⁰ thalassemia
- Hb D/Beta⁺ thalassemia
- Hb Variant/Beta⁰ thalassemia
- Hb Variant/Beta⁺ thalassemia
- other hemoglobinopathies (Hb variants)

* Actual start date of Program expansion to be announced via letter.



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